

Exon-Intron Boundary Sequences of the Human alpha-7 nAChR Subunit Gene

Exon Number	Exon length (bp)	CDNA position	splice acceptor	flanking exon sequence	splice donor	Intron number	Intron Size approximate (Kb)
1	55	1-55			...CTG CAC G L H	1	0.3
2	140	56-195	TCTCCCTTAAG	TG TCC . . . GAC GTG	GTGAGTCCCC	2	Unknown
3	45	196-240	TTTTTGAAAG	V S . . . D V			
4	110	241-350	TGTGTGTCAAG	GAT GAG . . . CAA ATG	GTAAGTTAAC	3	9.0
5	80	351-430	CTGTTCTAG T GCT GAT . . . CCT CCA G	Q M S W . . . N S	GTAAGCATAT	4	Unknown
6	168	431-598	ACCCACACAG	TCT TGG . . . AAC AG	GTAAGCTGCA	5	4.0
7	195	599-793	CCCTATGGAG	A D . . . P P			
8	87	794-880	TATGTTTAG	GC ATA . . . CTA GTG G	GTAAGCCATG	6	1.0
9	110	881-990	CTCTCCACAG	G I . . . L V			
10	519	991-1509	GTCTCCCCAG	GA ATC . . . TCC CTG G	GTAAGGGCCC	7	1.0
				G I . . . S L			
				GG ATA . . . TTG ATA G	GTAAGGCAAG	8	3.5
				G I . . . L I			
				CC CAG . . . AAG TGG	GTACGTTCT	9	5.0
				A Q . . . K W			
				ACC AGA . . .			
				T R			

FIG. 1

Sequence Variants Identified in Full-Length and Duplicated Genomic Clones

DNA	EXONS CONT.	EXON 6 +/- 497-498		EXON 7 654		EXON 7 690		EXON 10 1269		EXON 10 1335		L76630
		T/G	A/T	C/T	G/A	C/C	T/T	C/C	C/C	C/C	C/C	
CHR15 HYBRID	5-10 1-10	+TG -TG										6GT 8GT
YAC												
D-948a10	5-10	-TG						A	C			6GT
D-853b12	6-10	-TG				T		A	C			6GT
D/F 969b11	5-10 1-10	+TG -TG			C/T			A	C			6GT
F-134h10	1-10	+TG		C			G/A					8GT
F-776a12	1-10	+TG		C					C			8GT
F-791e6	1-10	+TG		C			G		C			8GT
F-811b6	1-10	+TG		C			G		C			8GT
F-953g6	1-10	+TG		C			G		C			8GT
F-859c11	1-10	+TG		C			G		C			8GT
F-810f11	1-10	+TG		C			G		C			8GT
F-801e1	1-10	+TG		C			G		C			8GT
BAC												
F-467o18	1-10	+TG		C			G		C			T
												8GT

FIG. 2

DNA	Control #	EXON 6		EXON 7		EXON 7		EXON 10		EXON 10		1335
		T/G	A/T	C/C	T/T	G/G	G/A	A/A	C/C	C/T	T/T	
Control Genomic DNA	43	+/- 10	+/- 33	-/- 0	C/C 5	T/T 38	G/G 0	G/A 43	A/A 0	C/C 6	C/T 36	T/T 1

Expression Analysis of Sequence Variants

	Bases 497-498			Base 654			Base 690			Base 933			Base 1296			Base 1335		
Subj	DNA	1-10 cDNA	5-10 cDNA	DNA	1-10 cDNA	5-10 cDNA	DNA	1-10 cDNA	5-10 cDNA	DNA	1-10 cDNA	5-10 cDNA	DNA	1-10 cDNA	5-10 cDNA	DNA	1-10 cDNA	5-10 cDNA
SL061	+-	+	+-	CT	C	CT	GA	G	GA	G	G	G	CT	CT	CT	C	C	C
SL084	+	+	+	C	C	C	GA	G	GA	G	G	G	CT	CT	CT	C	C	C
SL111	+-	+	+-	CT	C	CT	GA	G	GA	G	G	G	CT	CT	CT	CT	CT	CT
SL097	+	+	+	CT	C	CT	GA	G	GA	G	G	G	CT	CT	CT	C	C	C
SL089	+	+	+	C	C	C	GA	GA	GA	GA	GA	GA	CT	CT	CT	C	C	C
SHSY	+-	+	+-	CT	C	CT	GA	GA	GA	GA	GA	GA	C	C	C	C	C	C

FIG. 3

-392	agaacgcaag ggagaggtag agccctggcct tggcag <u>ccc</u> ctggactggc cagaggcg cagggcaggag
-322	cccgctcggt ggagactggg ggtggagggtg cccggaggct acccaggcgt ggaggatccct cccggctcaca
-252	cctcggtcg cagtccctg ggtggccggc gagacgcgtgg cccgggtcg aggatggg gggggggac
-182	gggggggg gcggggctcg tcacgtggag aggccgggg gggggggg ggccgggg Sp1
	CREB
-112	tccattaaagg cgccggagg gaggccggat gtggctctgt ggccggagg gcaaggccgg gggacacgg
-42	agacgtggag cgccggggct cgctgcggct ccgtcaatcg actATGGGCTG CTCCGGGGAA GGCCTCTGGC
+29	Met TGGCGCTGGC CGCGTCGCTC CTGGCACGGTA aagccac

FIG. 4

CENTROMERE			ALPHA-7 SEQUENCE										TELOMERE											
PAC	BAC	SIZE kb	D15S995					D15S1007					D15S144					D15S1010						
			-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	
PAC			-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-		
64a1			-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-		
25919			-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-		
BAC			-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-		
467	18		-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-		
YAC			+ 948a10	+ 853b12	+ 895f6	+ 969b11	+ 776a12	+ 791e6	+ 811b6	+ 953g6	+ 134h10	N.A.	+ 859c11	+ 810f11	+ 801e1	+ 966a4	+ 764f8	+ 822g2	+ 1330	+ 940	+ 1630	+ 1500	+ 740	+ 1280
			1730	790	1580	1030	1640	1170	1060	1720														

FIG. 5

1	CAGGGCCCA CACGGGGCG AGGGGCTCG TTCAAGGCCA CCCACGGCAA	CATAGCTCCC GGGAGGCTG GACTGGCTCC AACCGAAGRT CATTAAAGgtg	GCCAAGTCCCT GAGCCGGGAG TTTTCGGCGC ACTGGCCTCT AGtgcggcc.....	CGGTGCCCT AGCTGGCCG CCCTCCGGCC GGAGGTCAAG GGAACCAGGA	TGCCATTTC GGGGCCCCG CCCTCCGGC GGAAAGATGTC GCCACAGGCC	CAGCCGGTCTC CTGGTGGCCG GCAGGATGTC GCCACAGGCC	CCACGAGGGT CGGCCATGAC CATCTCAGGC CGGCTCACGC
EXON D 297bpctc TATCTACACG AAGtgtggtag	atttcgATT ACTCAGATCT ta.....	298 ACAAGTGGAC TGTGTCAAC ta.....	ACCTGAGTCA CCCATTATTG	GCAGGACCTG ACAATCCAAA	GAATCCAGA GGTGCAGAAA	TGAGAGAGCT GCACTCTGAC
EXON C 1125bptt TTTCAGgttag	tttaaccac 486 aca.....	423 agATAATGAA gatcat.....	ACAACCACCA 487 AATTGCTTAAT	TCGGTTAAAT CCAGGATTG	TTGATGCCAA TGGATAGCTG	AATATTGCAT CTACCAAGCAT
EXON B 64bpttta aca.....						
EXON A 47bpttta aca.....						
EXON 5 80bpctgtttc	tagtgcgtat GCATTGCCAG	534 GAGGCACATT TACCTGCCTC	ACGCCACATT CAGgttaagctgca..... 613	CCACACTAAC	GTGTGTTGCA	ATTCTCTGCG 533
EXON 6 27bpacccaca	caggCATATT	614 CAAGACTTCCC	TCCTACATCG	640	

FIG. 6

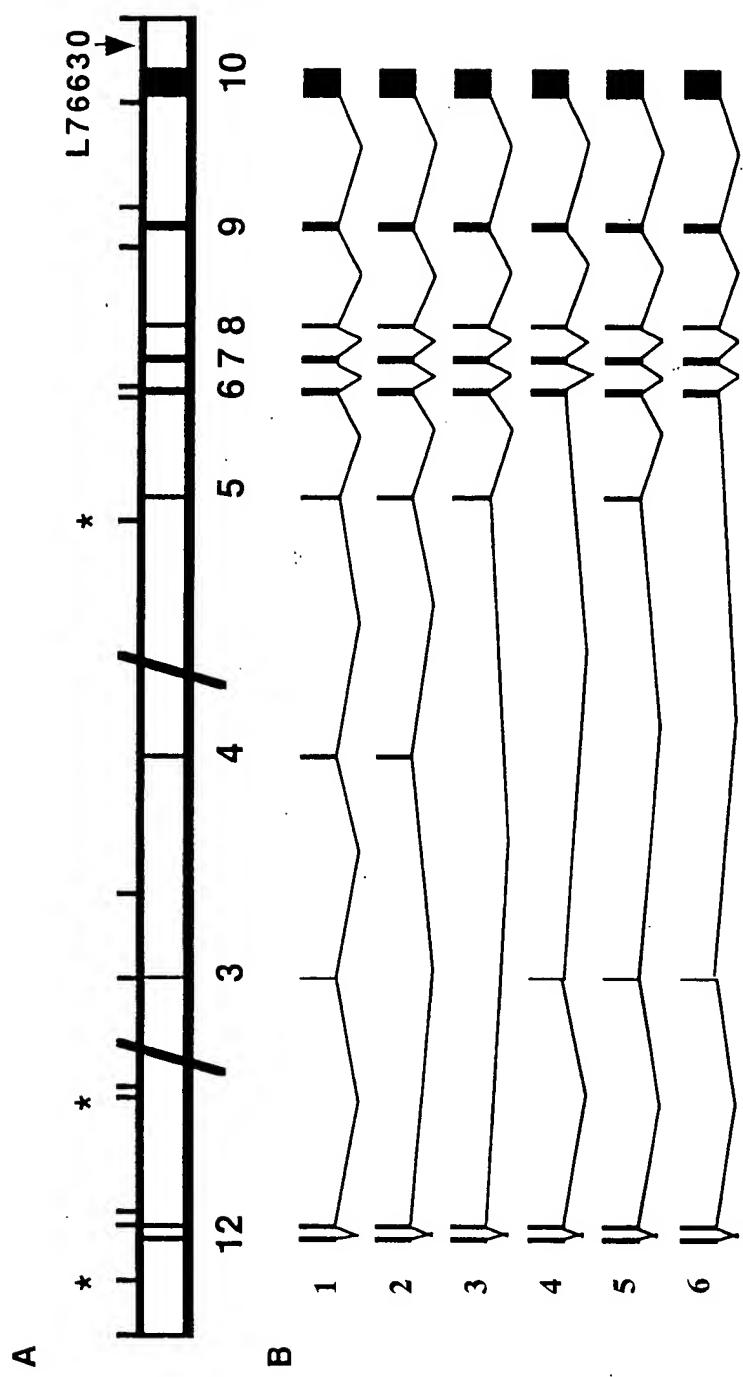


FIG. 7

1 agaacgcaag ggagaggtag agcctggcct tgggcagccc ctggcctggc cagaggcgcg
61 aggccgagag cccgctcggt ggagactggg ggtggaggtg cccggagcgt acccagcgcc
121 gggagtagacct cccgctcaca cctcgggctg cagttccctg ggtggccgccc gagacgctgg
181 cccgggctgg agggatggcg gggcggggac gggggcgaaa gcggggctcg tcacgtggag
241 aggcgcgcgg gggcgggcgg ggcgggggcg cgcgcggc tccttaaagg cgccgcgagcc
301 gagcggcgag gtgcctctgt ggccgcaggc gcaggccgg ggcacagccg agacgtggag
361 cgccggcggct cgctgcagct ccggactca ac

FIG. 8

1 caggccgcca catagctccc gccaagtccct cggtgccctt tgccattttc cagccgcgt
61 cccacgaggg tcacggcggc ggggagaggt ggagccgcga gagtcggcc gggggccccg
121 cctggggcc gcggccatga cagcggtcg ggactggctc ctttccgcg cccctccgc
181 cggaggtgag gggaaagatgt ccatgtcagg gttcaaggcc aaaccgaagt tactggcctc
241 tatctccag gagaaccagg agccacagcc gcggctcacg ccccacccga acattaagat
301 tacaagtggc cacctgagtc agcaggaccc ggaatccag atgagagagc ttatctacac
361 gactcagatc ttgttgtcac cccattatt gacaatccaa aggtgcagaa agcactctga
421 caaataatga aacaaccacc atcggttaaa ttgtatgcaa aaatattgca tctaccagca
481 tttcagttc caattgctaa tccagcattt gtggatacgct gcaactgcg atattgctga
541 tgagcgcttt gacgccacat tccacactaa cgtgttggtg aattcttctg ggcattgcca
601 gtacctgcct ccaggcatat tcaagagttc ctgctacatc g

FIG. 9

1 agccctttcc caggcggtag cgggggcagt ggtgctgttgc ccctttaaa ctgcggcttg
61 acgggagccg cgcctcctgt cggtggagtc gggtataaaag ggagcagccc cgcaggccgc
121 cacatagctc cccccaagtc ctcgggtcccc cttgccattt tccagccgcg ctcccacgag
181 ggtcacggcg gcggggagag gtggagccgc gagagctcg ccgggggccc cgcctggtgg
241 cccgcggccat gacagcggtc cgggactggc tcctttccg cgccctccc gccggaggtg
301 agggaaagat gtccatgtca gggttcaagg ccaaaccgaa gttactggcc tctatctcc
361 aggagaacca ggagccacag ccgcggctca cggccaccg caacattaag attacaagtg
421 gacacctgag tcagcaggac ctggaatccc agatgagaga gcttatctac acgactcaga
481 tcttgggtgc accccatata ttgacaatcc aaaggtgcag aaagcactct gacaattcca
541 attgctaattc cagcatttgt ggatacgctgc aaactgcgat attgctgatg agcgcttga
601 cgccacatcc cacactaacg tgggtgtgaa ttcttctggg cattgccagt acctgcctcc
661 aggcatattc aagagttc gctacatcg

FIG. 10

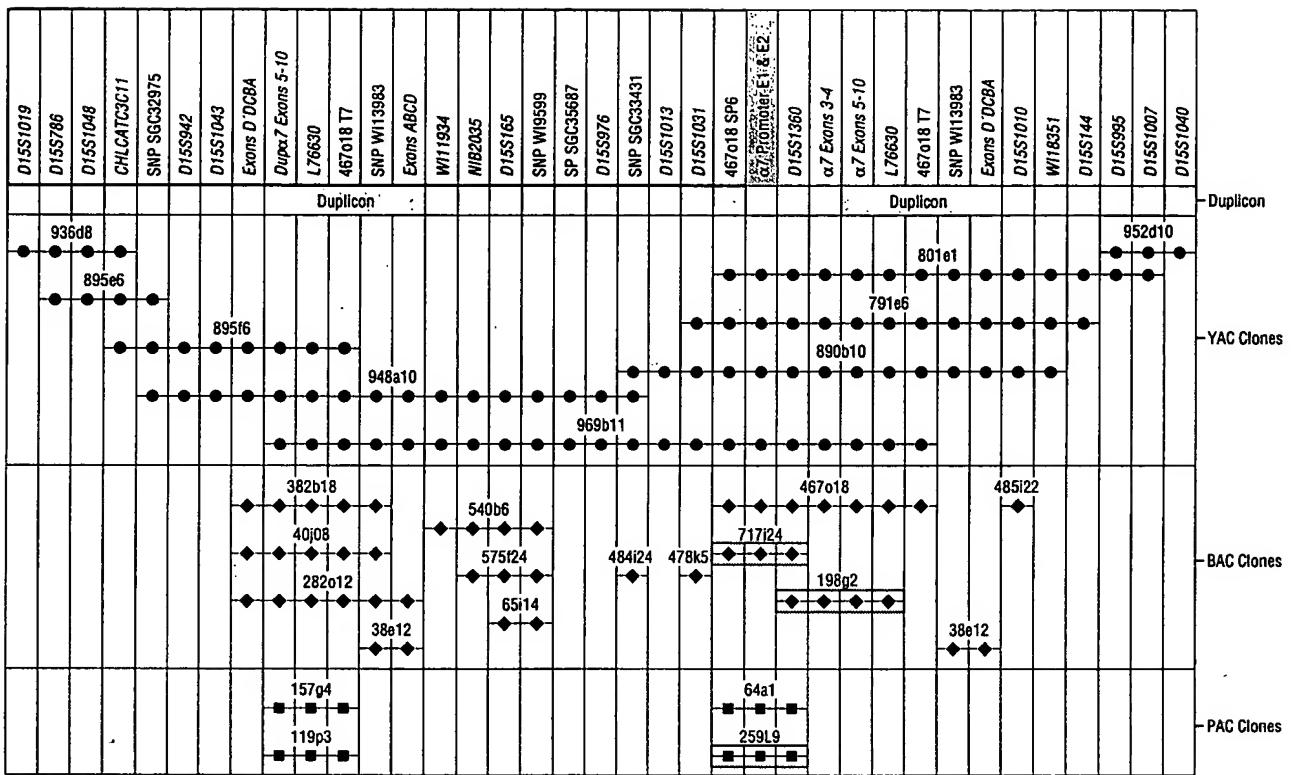


FIG. 11

A



B

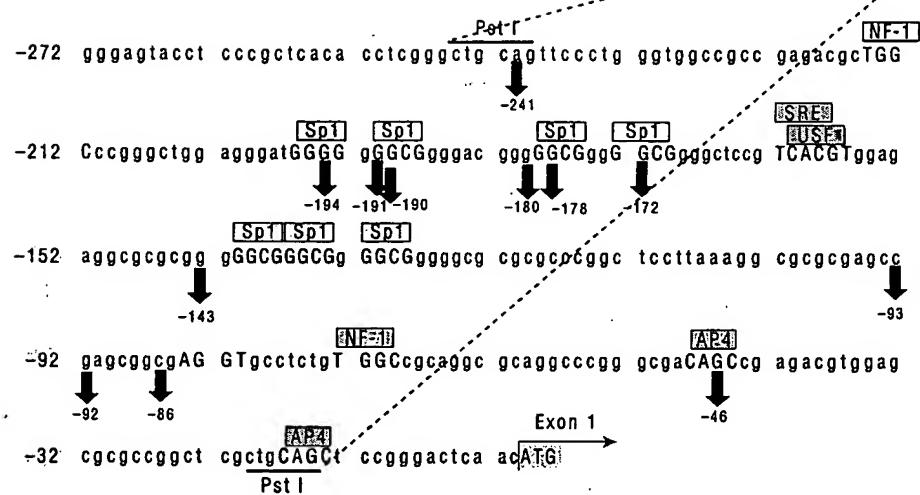


FIG. 12

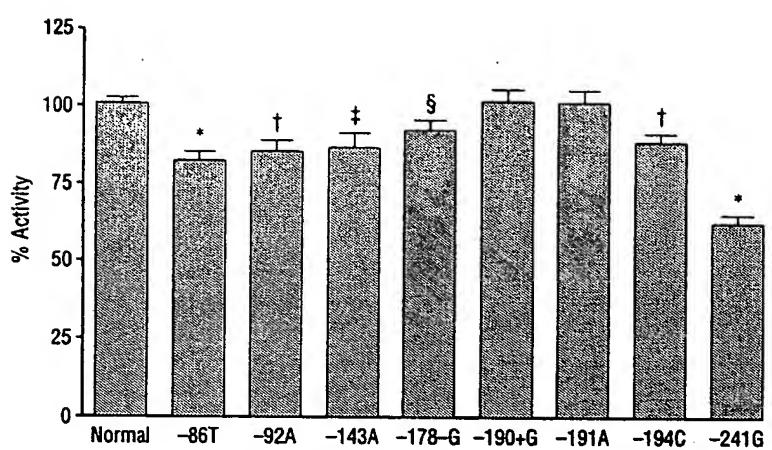


FIG. 13

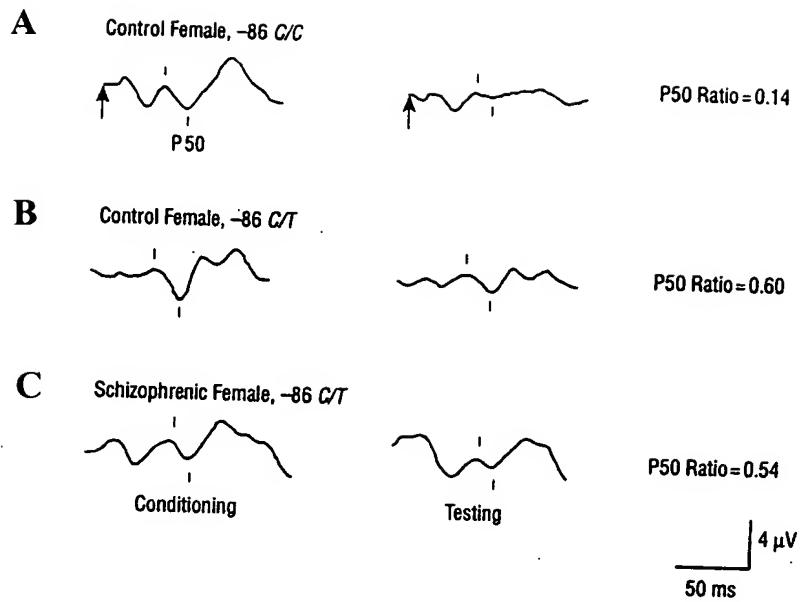


FIG. 14

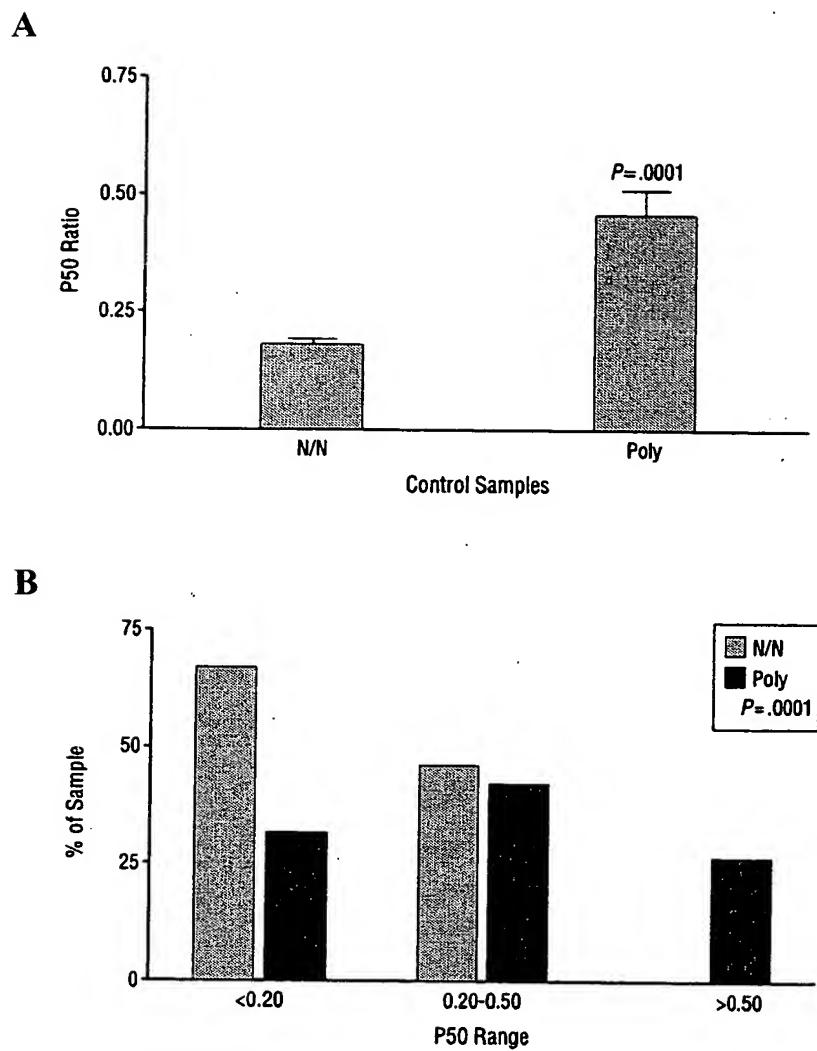
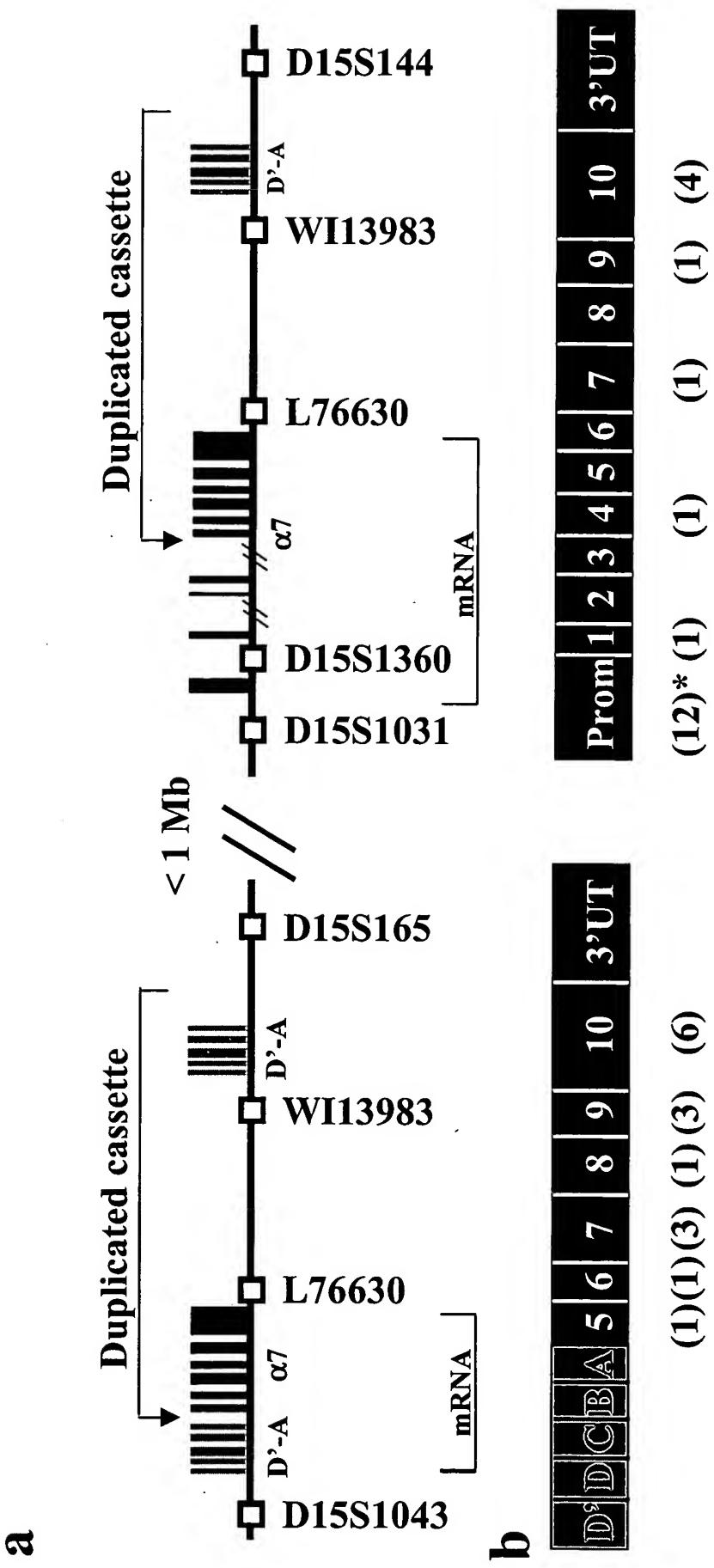


FIG. 15

FIG. 16



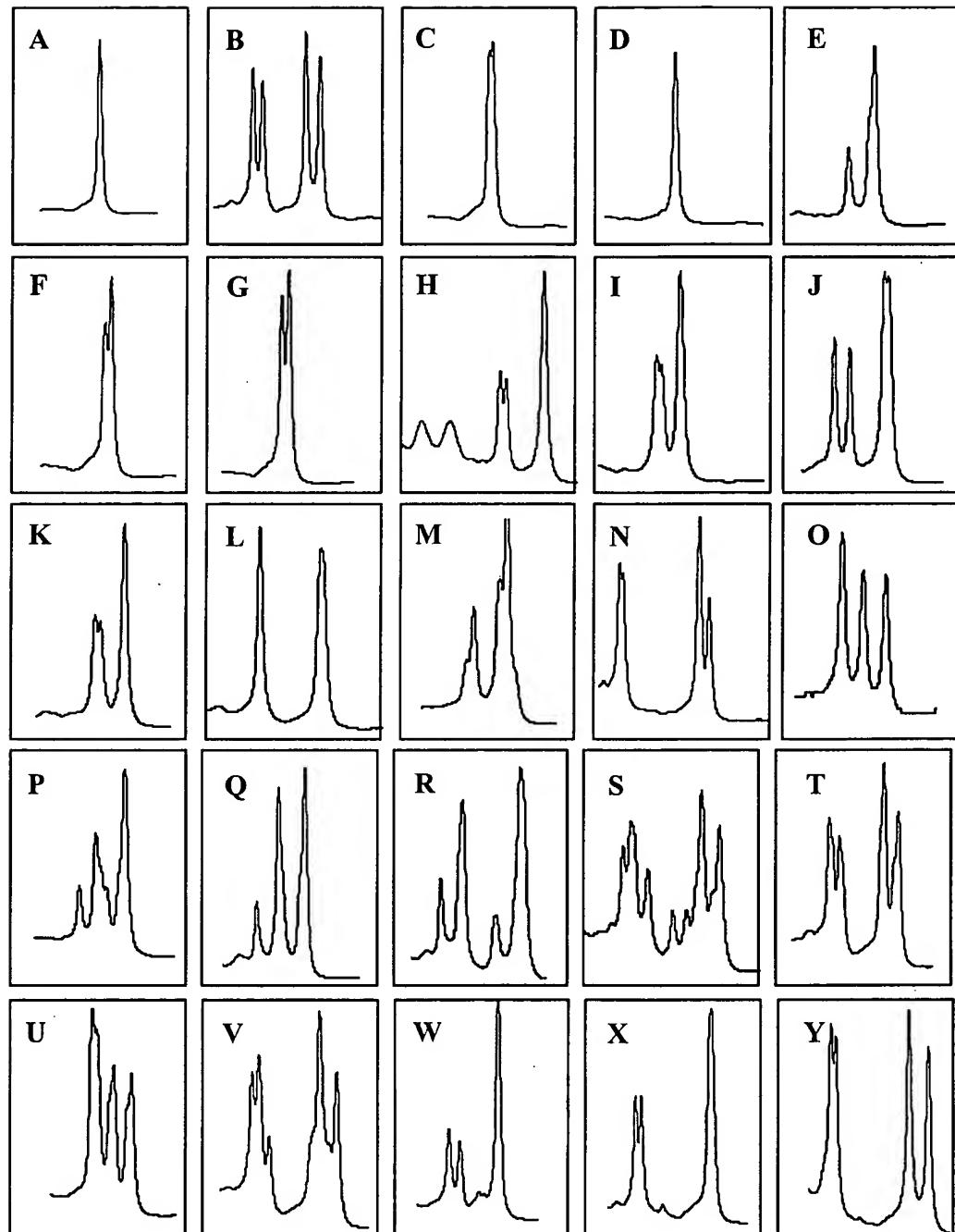


Fig. 17